Speaker: Dorothy Kim Waller, Ph.D.

Topic: Obesity, Diabetes and Birth Defects

Objectives:

1) Summarize the literature regarding obesity as a risk factor for birth defects.
2) Summarize the literature regarding poor glycemic control and different categories of diabetes as risk factors for birth defects.
3) Describe how obesity and diabetes are related.

Outline:

1) Description of the major types of diabetes
   a) Insulin-dependent diabetes mellitus (IDDM)
   b) Non-insulin dependent diabetes mellitus (NIDDM)
   c) Gestational diabetes
2) Increased risk of offspring with major congenital malformations among women with insulin-dependent diabetes mellitus (IDDM)
3) Increased risk of offspring with major congenital malformations among pregnant mice and rats with artificially induced IDDM
4) Good glycemic control prior to conception reduces the risk of offspring with malformations in both humans and in animals
5) Types of malformations increased among women with IDDM
6) Little or no increase in risk of offspring with congenital malformations among women with gestational diabetes
7) Increase in risk of offspring with neural tube defects, especially spina bifida among non-diabetic obese women
8) Characteristics of obese women suggesting that they have a higher prevalence of *pre-clinical diabetes*
   a) Increased risk of gestational diabetes
   b) Increased risk of developing NIDDM in middle-age
   c) Higher serum levels of insulin
   d) Increased prevalence of marosomic infants
9) Abdominal obesity versus gynoid obesity
10) Association between particular congenital malformations and macrosomia
11) Other explanations for an association between obesity and congenital malformations

Speaker: Lori Wolfe, M.S., CGC

Topic: The Texas Teratogen Information Service

Objectives:
1) Define the term Ateratogen@
2) Give three examples of common teratogens.
3) Describe the purpose and target audiences of the Texas Teratogen Information Service.
4) Give one example each of how this service has helped a provider and a parent.

Outline:
1) Introduction
   a) View 90 second PSA
2) Slide presentation
   a) Define teratogen
   b) Teach 3 stages of pregnancy and different teratogenic risks
   c) Review common teratogens and the related pregnancy risks
   d) Present teratogen statistics for the state of Texas

Abstract: The Texas Teratogen Information Service exists to provide the residents of Texas with information regarding pregnancy exposures and related risks. Any exposure that occurs during pregnancy and increases the risk for birth defects, miscarriage, and/or learning problems is considered to be a teratogen. Factors determining possible increased risk include the timing in the pregnancy of the exposure, the dosage used, the route of the exposure, and any positive health and genetic histories of the exposed individual. Common teratogenic agents include alcohol, antidepressants and other prescription medications, cigarette smoking, non-prescription medications, cocaine and other street drugs, and occupational and environmental exposures.

The Texas Teratogen Information Service provides teratogen information and counseling, free of charge, to thousands of individuals and health care professionals each year. In addition, the service has many handouts that are available to educators and health care professionals to use as needed.

Speaker: Lowell Sever, Ph.D.

Topic: Latest Developments in the Environmental Causes of Birth Defects

Objectives:

1) Name three recent studies published about birth defects and the environment.
2) Name three types of environmental factors that are being researched, and their suspected effects.
3) Give an example of how knowledge of environmental causes can translate into prevention activities.
4) Name two environmental effects of special interest to Texas, and how they are geographically associated.

Outline:

1) Agents and exposure sources of particular current concern
   a) Water disinfection byproducts
   b) Pesticides and other agricultural chemicals
   c) Hazardous waste sites
2) Assessment of exposure
   a) Hierarchy of exposure assessment approaches
   b) Exposure pathways
   c) Timing of exposure
   d) Heterogeneity of exposures: dealing with mixtures
3) Lumping and splitting: the importance of how defects are grouped
   a) Neural tube defects
   b) Oral facial clefts
   c) Heart defects

Abstract: There are many questions and concerns about potential associations between environmental agents and risks of birth defects in humans. To a large degree, the information we have regarding risks is not convincing. At the same time, there is increasing evidence suggesting that some environmental contaminants are associated with reproductive or developmental toxicity.

In this presentation I will review some of the recent studies relating to environmental contaminants and the etiology of congenital malformations. I will consider three general categories of exposures: water disinfection byproducts, pesticides and other agricultural chemicals, and hazardous waste sites. In reviewing the evidence for developmental toxicity associated with these exposures, I will look particularly at how exposure has been assessed and how defects have been grouped. Key issues relative to exposure assessment include having a completed exposure pathway and the timing of exposure. In discussing the grouping of outcomes, I will focus on neural tube defects, oral facial clefts, and heart defects to illustrate some of the major problems that have been encountered in studies to date and how they have been, or can be, addressed.

Speaker: Joanna Spahis, R.N.

Topic: Perinatal Issues in Down Syndrome

Objectives:

1) Introduction
2) Patients and Methods
   a) Gestational Information
   b) Prenatal Diagnosis
   c) Neonatal Information
3) Neonatal Complications
   a) Gastrointestinal Anomalies
   b) Other Gastrointestinal Problems/Feeding Difficulties
   c) Cardiac Anomalies
4) Counseling Experiences
   a) How Parents Were Informed
   b) Parents= Impressions of Initial Diagnosis Experience
   c) Limitations of Study
5) Recommendations
   a) Jaundice
   b) Feeding
   c) Constipation/Anal Stenosis
   d) Initial Counseling
   e) Miscellaneous

Abstract: This presentation is a summary of a study done at Children=s Medical Center of Dallas and the University of Texas Southwestern Medical Center between 1993 and 1998. Its purpose was to identify perinatal complications among infants with Down syndrome and to explore the early counseling experiences of families with regard to this new diagnosis. The sample included 216 families (127 males and 89 females with DS) who completed medical and gestational history forms, supplemented with interviews by the clinical nurse specialist at their DS clinic visit. Data were recorded in a Microsoft Access 97 database and queried to establish frequencies of clinical findings and other responses. Results of prenatal screening measures are given. At birth, 94% of infants were suspected of having DS and 80% of them were karyotyped. Approximately 51% of these infants had echocardiograms prior to discharge. There was a high incidence of gastrointestinal problems reported (77%), mostly from physiologic jaundice and feeding issues. Cardiac defects were also prevalent in 38% of infants. A summary is given from the early counseling experiences of families. Recommendations are made for the most common clinical findings as well as counseling strategies that will benefit families with a suspected diagnosis of DS.

Texas Birth Defects Conference 2000
Perinatal Issues in Down Syndrome
Joanna Spahis, MSN, RN

Goals of the Study:

- To identify less recognized neonatal problems in the DS population
- To validate and update the Preventive Medical Checklist for DS based on new findings
- To explore early counseling experiences of families with regard to the new diagnosis of DS
- To increase the awareness of positive strategies for health care practitioners when giving a new diagnosis of DS to a family

Gestational Findings:

- 58 mothers (27%) over age 35 years
- 19% of infants reported to be born at < 37 weeks gestation
- Low birth weight not common
- 9% weighed < 3rd centile for gestational age
- Mean birth weight = 3.0 kg

Prenatal Diagnosis Findings:

- Amniocentesis or MSAFP screening reported in 83 pregnancies (39%)
- 31 results recalled as "abnormal" (37%)
- 12 of 31 women with "abnormal" results declined further testing (39%)
- 22 women received definitive prenatal diagnosis of DS by amniocentesis with or without prior MSAFP (10%)

Neonatal Management Findings:

- 200/216 infants (94%) suspected of DS at birth
- 169/200 infants (80%) karyotyped at birth
- 108/200 infants (51%) had echocardiograms prior to discharge

Perinatal Issues in Down Syndrome

Joanna Spahis, MSN, RN, Golder N. Wilson, MD, PhD

Children's Medical Center of Dallas and The University of Texas Southwestern Medical Center

Introduction

- Certain neonatal complications in DS are well known, including:
- Cardiac malformations (40-50%)
- Gastrointestinal anomalies (12%)
- Hematologic abnormalities (5-10%)

Goals of the Study

- To identify less recognized neonatal problems in the DS population
- To validate and update the Preventive Medical Checklist for DS based on new findings

Additional Goals...

- To explore early counseling experiences of families with regard to the new diagnosis of DS
- To increase the awareness of positive strategies for health care practitioners when giving a new diagnosis of DS to a family

Patients and Methods

- Medical evaluations performed on 669 outpatients with DS (930 visits) seen in a weekly, multidisciplinary DS clinic between May 1993 and January 1998
- Medical and gestational history forms completed by families, supplemented with interviews by clinical nurse specialist

Patients & Methods

- 216 patient records were sufficiently recent (children < 2 yrs at visit) and complete for analysis
- Sample included 127 males and 89 females
- Data recorded in Microsoft Access database and queried to establish frequencies of positive responses or clinical findings

Gestational Findings

- 58 mothers (27%) over age 35 years
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Neonatal Management Findings

- 200/216 infants (94%) suspected of having DS at birth
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- 108/200 infants (51%) had echocardiograms prior to discharge

Gastrointestinal anomalies N = 37 (17%)

Anal stenosis	N = 24 (11%)
Duodenal stenosis/atresia	N = 10 (5%)
Imperforate anus	N = 2[A] (1%)
Hirschsprung disease	N = 2 (1%)

A = Fewer than 216 patients or responses

Gastrointestinal problems N = 166 (77%)

Feeding problem (all types)	N = 123[A] (57%)
Feeding problem (bottle only)	N = 61/95[A] (64%)
Feeding problem (breast / bottle)	N = 38/67[A] (57%)
Jaundice (no phototherapy)	N = 147 (68%)
Jaundice (with phototherapy)	N = 73 (34%)
Constipation	N = 40 (18%)

A = Fewer than 216 patients or responses

Factors contributing to feeding difficulties

- Hypotonia
- Poor suck/swallow coordination
- Complications of cardiac disease
- Prematurity
- Unspecified infant fatigue/lethargy

Cardiac anomalies N = 82 (38%)

Ventricular septal defect	N = 25 (12%)
Atrial septal defect	N = 19 (9%)
Atrioventricular septal defect	N = 12 (6%)
Patent ductus arteriosis	N = 16 (8%)
Tetralogy of Fallot	N = 7 (3%)

Counseling Issues: How parents were informed

Informed by physician	188 (87%)
Informed by other health professional	28 (13%)

Who was there at the time of diagnosis?

Both parents present	N = 105/178[A] (59%)
One parent present	N = 73/178[A] (41%)

A = Fewer than 216 patients or responses

Overall impression of initial diagnosis experience

Positive impression	N = 66 (31%)
Neutral/mixed impression	N = 66 (31%)
Negative impression	N = 83 (38%)

Reasons Given for Positive Experience

- Positive attitude of physician
- Presence of spouse when told
- Time given to recover from delivery
- Privacy to grieve
- Provision of accurate information re: DS
- Provision of additional resources

Reasons Given for Negative Experience

- Negative, rude, or abrupt manner of physician
- Information given too soon after birth
- Spouse absent
- Infant not able to be viewed
- Inaccurate or insufficient information about DS

Limitations of Study

- Sample drawn from only one region in North Texas
- Bias of parental recall
- Neonatal records unavailable in some cases to supplement parent questionnaires and interviews

Recommendations: Jaundice

- Parent education regarding signs and symptoms of jaundice
- Parent education regarding frequent feedings, assessment of stools
- Follow-up for infants discharged routinely

Recommendations: Feeding

- Lactation support for mothers who wish to breastfeed
- Referral to early childhood intervention program for therapy to improve hypotonia
- Feeding assessment by speech therapist and video swallow if needed

Recommendations: Constipation, Anal Stenosis

- Obtain history: frequency of bowel movements, pain, straining at BM's
- Rectal exam to confirm anal stenosis
- For constipation, use Milk of Magnesia 1/2 cc/kg daily
- For anal stenosis, instruct parents in gentle manual rectal dilation

Recommendations: Initial Counseling

- Emphasize positive aspects of infants with DS
- Wait for spouse or support person to give news
- Allow parents time to view and bond with baby and recover from delivery
- Give information in a private location
- Provide information/referrals for local resources (ECI, DS Clinic, support group)

Additional Recommendations

- Use of Down Syndrome Preventive Medical Checklist to address multiple medical, developmental, and psycho-social issues
- Use of Down Syndrome Growth Charts to monitor length and weight gain

Medical Complications at Birth:

- Congenital gastrointestinal defects (anal stenosis, duodenal stenosis/atresia, imperforate anus, Hirschsprung disease) = 17%
- Gastrointestinal problems (feeding issues, jaundice, constipation) = 77%
- Congenital cardiac defects (VSD, ASD, AVSD, PDA, Tetralogy of Fallot) = 38%

Counseling Issues:

- Parents informed by physician in 87% of cases, by other professional in 13% of cases
- Parents informed together in 59% of cases, mother informed alone in 41% of cases
- Overall impression of initial counseling experience: positive (31%), neutral/mixed (31%),
  negative (38%)

Recommendations: Jaundice

- Parent education regarding signs and symptoms of jaundice
- Parent education regarding frequent feedings, assessment of stools
- Home follow-up for infants after routine discharge

Recommendations: Feeding

- Lactation support for mothers who wish to breastfeed
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- Use of Down Syndrome Preventive Medical Checklist to address multiple medical, developmental, and psycho-social issues
- Use of Down Syndrome Growth Charts to monitor length and weight gain

Speaker: Wendy Nembhard, MPH

Topic: Survival Patterns for Infants with Birth Defects in Texas

Objectives:

1) Name the two birth defect categories with the best and poorest survival rates during infancy.
2) Describe the regional survival patterns for one birth defect category.
3) Identify a significant racial/ethnic or socioeconomic pattern seen in the survival patterns.

Outline:

1) Introduction
    a) Infant Mortality in United States 1960-1999
    b) Infant Mortality among infants with birth defects in Texas
2) Description of Study
    a) Study Design
    b) Description of Study Population
    c) Methods
    d) Description of two birth defects
3) Presentation of Results
    a) Explanation of Calculations
        i) Infant Mortality Rate
        ii) Kaplan-Meier Survival Curves
    b) Birth Defect with Best Survival Rate
        i) Infant Mortality Rate
        ii) Kaplan-Meier Survival Curve
    c) Birth Defect with Worst Survival Rate
        i) Infant Mortality Rate
        ii) Kaplan-Meier Survival Curve
    d) Regional Pattern for Birth Defect
        i) Infant Mortality Rates
        ii) Kaplan-Meier Survival Curves
    e) Racial/Ethnic Differences in Survival for Birth Defect
        i) Infant Mortality Rates
        ii) Kaplan-Meier Survival Curves
4) Discussion of Results
    a) Birth Defect with Best Survival
    b) Birth Defect with Worst Survival
    c) Regional Patterns
    d) Racial/Ethnic Differences
5) Limitations of Study
6) Future Research
7) Conclusion

Speaker: Karla Damus, R.N., Ph.D.

Topic: Planning for the Unplanned Pregnancy: Primary Prevention of Birth Defects

Objectives:

1) Define the term Apreconceptional care@
2) Name to barriers to the provision of preconception planning.
3) Identify three birth defects which could be prevented through preconception planning.

Outline:

1) Preconceptional care is the cornerstone of primary prevention of adverse outcomes of pregnancy
2) Risk factors associated with higher incidence of birth defects are increasing in the US and Texas including advanced maternal age, plurality, pre-existing chronic conditions (such as diabetes, hypertension, autoimmune disorders), and use of substances, especially alcoholic beverages and illicit drugs, in women of reproductive age
3) Although the cause of 2 out of 3 birth defects remains unknown, substantial primary prevention of some serious birth defects could be achieved with preconceptional and prenatal avoidance of known teratogens (e.g. alcohol, illicit drugs, certain medications, x-ray, etc), control of pre-existing medical conditions, and daily consumption of 400 mcg of folic acid
4) At least 50% of serious neural tube defects (spina bifida, anencephaly, encephalocoele) and a proportion of other birth defects affecting the spine and lower extremities, some outflow defects of the heart, urinary tract, cleft lip and palate and possibly Down Syndrome could be prevented by preconceptual care
5) There are many patient, provider, and system barriers to the provision of comprehensive preconceptional care involving the patient, the provider and the health care delivery system
6) It is cost effective to provide preconceptional care for many reasons including the prevention of some very serious birth defects